

GENOMICS: WHY SHOULD WE CARE?

The importance of being: genomic literate
genomic informed

Orphan Black



“We can provide you with a healthy and thriving newborn, but why stop there? All of our children are born stronger and healthier. At BrightBorn Technologies we’re making the world a better place, one baby at a time.”

News Headlines

Scientists Say They Hope To Create A
Human Genome In The Lab

Humans will be 'irrevocably altered' by genetic editing, warn scientists ahead of summit

An open letter from 150 scientists, campaigners and health experts is calling for a worldwide ban on genetic editing ahead of a summit in Washington

**The Genetic Tool That Will
Modify Humanity**

Crispr allows scientists to control the blueprints of life, for better or worse.

British Scientists Seek Permission To Edit
DNA In Human Embryos

**Clinical Genetics Has a Big
Problem That's Affecting
People's Lives**

Unreliable research can lead families to make health decisions they might regret.

Genetically Modified Humans?
How Genome Editing Works

Having a baby? Best to run a gene screening test

Birth of Baby With Three Parents' DNA Marks Success for Banned Technique



Health Literacy

Genomic Literacy



- Genomic health literacy
- Genomic science literacy
- Role of media in genomic literacy

Leading causes of death

1. Heart disease: 614,348
2. Cancer: 591,699
3. Chronic lower respiratory diseases: 147,101
4. Accidents (unintentional injuries): 136,053
5. Stroke (cerebrovascular diseases): 133,103
6. Alzheimer's disease: 93,541
7. Diabetes: 76,488
8. Influenza and pneumonia: 55,227
9. Nephritis, nephrotic syndrome, and nephrosis: 48,146
10. Intentional self-harm (suicide): 42,773

Definitions

- Genomic Health Literacy

- ▣ The capacity to obtain, process, understand, and use genomic information for health related decision making.

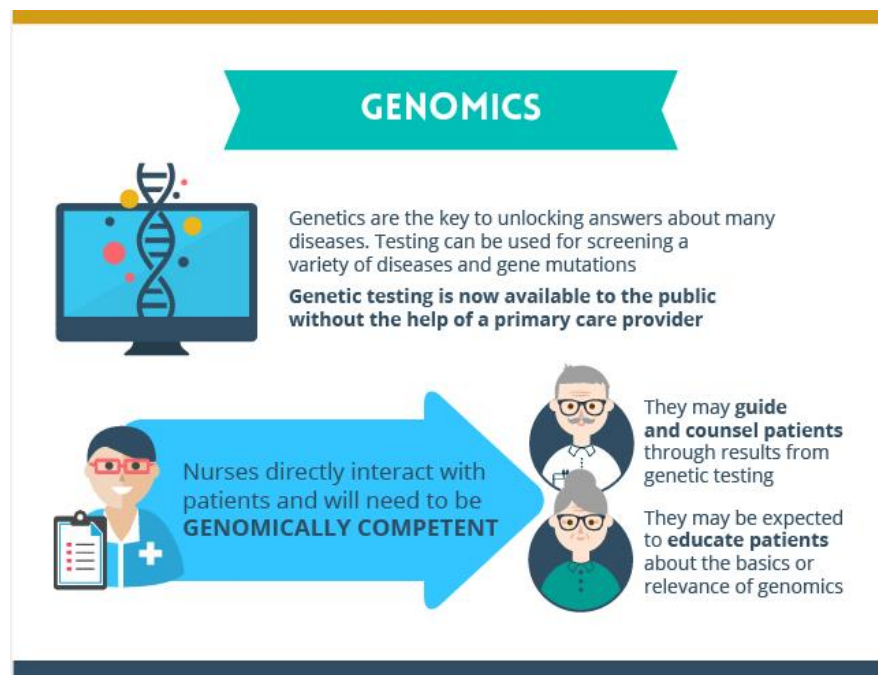
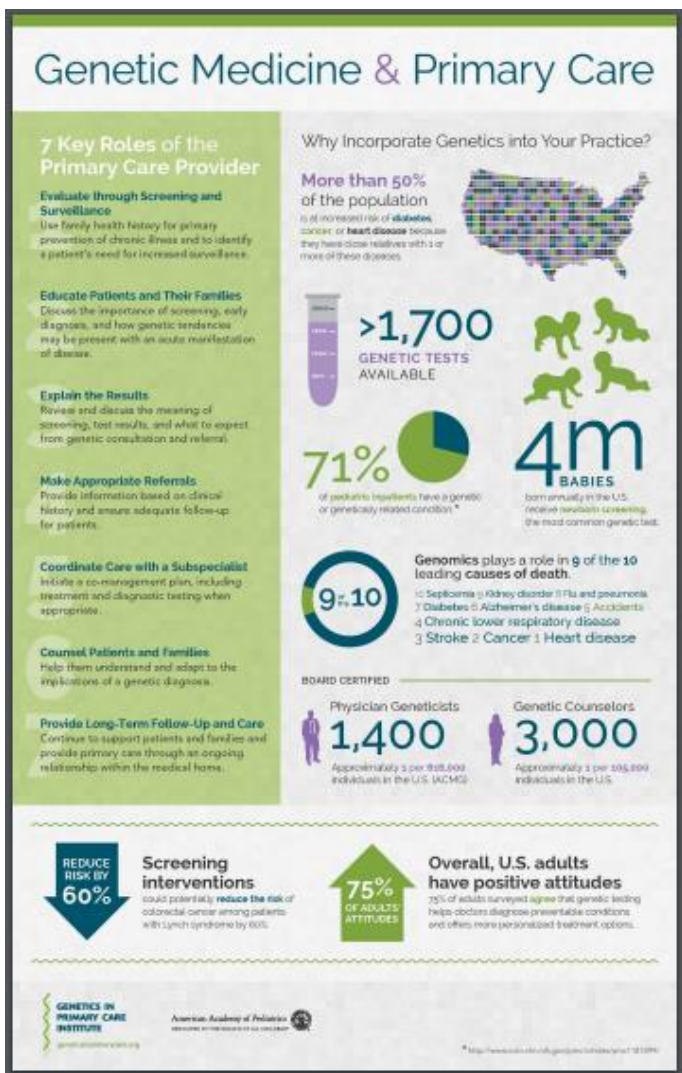
- Genomic Science Literacy

- ▣ The knowledge of basic genetics and genomics concepts and processes needed to build conceptual understanding, and the necessary mathematical knowledge to support this comprehension.

Health Professional Literacy

- Most have no formal training in genetics
- Many lack basic genomic knowledge
- Patients ill-informed, inflated expectations
- Lack of time to keep up to date with the genetic advances
- Uncertainty of relevance and how to incorporate in general practice
- Need to improve medical education and beyond initial training

Health Professional Literacy



Consumer Genomic Health Literacy

- ❑ Lack biology basics
- ❑ Lack mathematical concepts
- ❑ Low health literacy



Genomic Science Literacy

- ❑ K-12 education unable to keep up with scientific advancements
- ❑ Low emphasis on genomics
- ❑ Some teachers have misconceptions about genetics/genomics and little understanding
- ❑ Teachers need updated skills and have little access to genetic/genomic quality science curriculum
- ❑ Encourage partnerships with scientists
- ❑ Empower students entering the age of personal genomic medicine

Media role in genomic literacy

- ❑ Scientists lack training when communicating with media and general public
- ❑ Popular and mass media lack knowledge and often relay incorrect information
- ❑ Educators and researchers need to adapt and learn to inform through newer media platforms such as social media and podcasts



Educational Resources

NLM GeneEd

The screenshot shows the NLM GeneEd website homepage. At the top, there is a yellow banner with the GeneEd logo (a stylized DNA helix) and the tagline "Genetics, Education, Discovery". To the right of the logo, there are links for "Text Size +", "Print This Page", and "Email Us". Below the banner is a navigation bar with buttons for "Home", "Topics", "Labs & Experiments", "Teacher Resources", "Careers in Genetics", and "Highlights". A search bar is located on the right side of the navigation bar. The main content area is divided into a grid of topic cards. Each card features a small image, a title, and a brief description. The topics include Cell Biology, Genetic Conditions, DNA Forensics, DNA, Genes, Chromosomes, Evolution, Top Issues In Genetics, Heredity/Inheritance Patterns, Biostatistics, Epigenetics/Inheritance and the Environment, and Biotechnology. A large image of two students looking at a laptop with a DNA helix overlay is positioned on the right side of the main content area.

GeneEd *Genetics, Education, Discovery*

Text Size +
Print This Page
Email Us

Home Topics Labs & Experiments Teacher Resources Careers in Genetics Highlights search... GO

Cell Biology
The study of the cell, including mitosis and meiosis

Genetic Conditions
Conditions caused by gene variations or mutations

DNA Forensics
The application of genetic testing for legal purposes

DNA, Genes, Chromosomes
The building blocks of inheritance

Evolution
Processes by which organisms are changed over time

Top Issues In Genetics
Select genetic topics of popular interest

Heredity/Inheritance Patterns
The genetic transmission of traits

Biostatistics
The use of mathematics to investigate life science related problems

Epigenetics/Inheritance and the Environment
The genome's dynamic response to the environment

Biotechnology
Biological techniques used to enhance products

NIH National Human Genome Research Institute



Education

Educational materials about genetics and genomics



Smithsonian NHGRI Genome Exhibition
A genomics exhibition from the Smithsonian and NHGRI



About The Human Genome Project
Information on the history, progress and impact of the HGP



Talking Glossary of Genetic Terms
Terms and definitions used in genetic research with multimedia



Genetic Careers
Information on careers in genomics and genetics



National DNA Day
A unique day when everyone can learn more about genetics and genomics



Genetic Education Resources for Teachers
Teaching plans to present the science of genetics and genomics



Online Genetic Education Resources
A list of online resources for learning about genomics and genetics



Fact Sheets
Clearly written information on the institute, genetic research and genetic concepts



Online Education Kit
A web-based resource outlining the major history and developments of genomics

Highlights

Harry Potter and the Genetics of Wizarding



For NHGRI's National DNA Day inaugural speaker series, Eric P. Spana, Ph.D. from Duke University presented *Harry Potter and the Genetics of Wizarding*. The event took place on the NIH campus in Building 35A (Porter Building), Room 640. It is now available on NHGRI's YouTube channel, GenomeTV. Dr. Spana is an award-winning instructor in biology who helps students place new information in context with ideas they already find familiar, like Harry

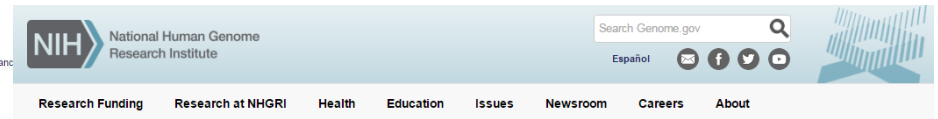
See Also

Education and Community Involvement Branch

GenomeTV

Genome Advance of the Month

Education Archive



Health

Information about genetics and genomics, rare diseases, patient care and more

For Patients and the Public



Detailed information about genetic disorders, background on genetic and genomic science, pharmacogenomics, family health history tool and online health resources

- › Community Engagement and Community Health
- › Family History
- › Genetics & Genomics Science & Research
- › Genetic & Rare Diseases Information Center
- › Genomic Medicine and Health Care
- › Online Health and Support Resources
- › Specific Genetic Disorders

For Health Professionals



Genetics and genomics information related to patient management, education, NIH and NHGRI research and ethical, legal and social issues

- › Competency & Curricular Resources
- › Genetics 101
- › Genomic Medicine and Health Care
- › Inter-Society Coordinating Committee (ISCC)
- › New Horizons and Research
- › Patient Management
- › Policy and Ethics Issues

Highlights

NIH awards \$55 million to build million-person precision medicine study



Bethesda, Md., Thurs., July 7, 2016 - The U.S. Food and Drug Administration (FDA) has announced two draft guidances to support President Obama's Precision Medicine Initiative. The guidances will help provide oversight for tests based on next generation sequencing, a technology that examines a person's DNA to detect medically important differences in genomic make-up that could increase the risk for disease.

See Also

GenomeTV

Genomic Healthcare Branch

Fact Sheets

Genetic Education Resources for Teachers

All About the Human Genome Project



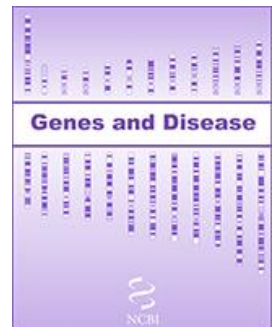
NCBI Bookshelf

Genes and Diseases

- ❑ Organized by the parts of the body that genetic disorders affect
- ❑ Over 80 genetic disorder summaries
- ❑ Images and interesting facts
- ❑ PDF downloads of chapters
- ❑ Links to related research literature and pertinent websites

Genes and Diseases

<http://www.ncbi.nlm.nih.gov/books/NBK22183/>



National Coalition for Health Professional Education in Genetics (NCHPEG)

The screenshot shows the NCHPEG website homepage. At the top is a red navigation bar with links: HOME, ABOUT NCHPEG, PRODUCTS AND PROGRAMS, ANNUAL MEETING, and NCHPEG EVENTS. Below the navigation bar are the NCHPEG logo (a stylized DNA helix) and The Jackson Laboratory logo (a stylized 'J' with a DNA helix). A search bar is located to the right of the logos. The main content area features a large graphic with the text 'HEALTH 2.0' in a cloud, surrounded by phrases like 'SHARING DATA FOR RESEARCH', 'COLLABORATION AND PRACTICE', 'STAYING INFORMED', 'MANAGING A PARTICULAR DISEASE', and 'MEDICAL EDUCATION'. To the right of this graphic is a sidebar with three sections: 'Collaborate' (with a 'Comment' icon), 'Educate' (with a 'Learn' icon), and 'Evaluate' (with a 'Good' icon). Below the main graphic is a 'PROGRAM HIGHLIGHTS' section with three featured programs: 'Medicine's Future: Genomics for Practicing Providers', 'GeneFacts', and 'Colorectal Cancer: Is Your Patient at High Risk?'. To the right of this section is a 'Pregnancy & Health Profile' section with a tree diagram and a 'DOWNLOAD HERE' button.

HOME ABOUT NCHPEG PRODUCTS AND PROGRAMS ANNUAL MEETING NCHPEG EVENTS

NCHPEG
National Coalition
for Health Professional
Education in Genetics

**The Jackson
Laboratory**
Leading the search
for tomorrow's cures

SHARING DATA
FOR RESEARCH

COLLABORATION
AND PRACTICE

STAYING
INFORMED

HEALTH 2.0

MANAGING
A PARTICULAR
DISEASE

Medicine's Future: A
Genomics Curriculum for
Clinicians

Engaging clinicians in continuing
education to integrate genomics
into th...

Read more

GeneFacts

GeneFacts is a point-of-care,
decision-support system for non-
geneticists...

Read more

Colorectal Cancer: Is Your
Patient at High Risk?

Learn to identify patients at
increased risk for hereditary
colorectal c...

Read more

Pregnancy & Health Profile

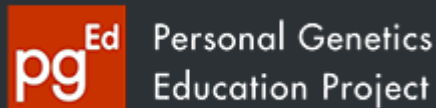
Genetic Screening, Risk Assessment
and Clinical Decision Support

DOWNLOAD
HERE

Literacy/Education Resources



Public Health Genomics



discover. educate. advocate.



Tips for communicating to patients

- Listen, pay attention, respond
- Use plain language
- Use patient's words
- Slow down
- Limit and repeat content
- Show examples
- Invited patient participating
- Use teach back

[Health Literacy and Precision Medicine](http://www.nationalacademies.org/hmd/Activities/PublicHealth/HealthLiteracy/2016-MAR-2.aspx)

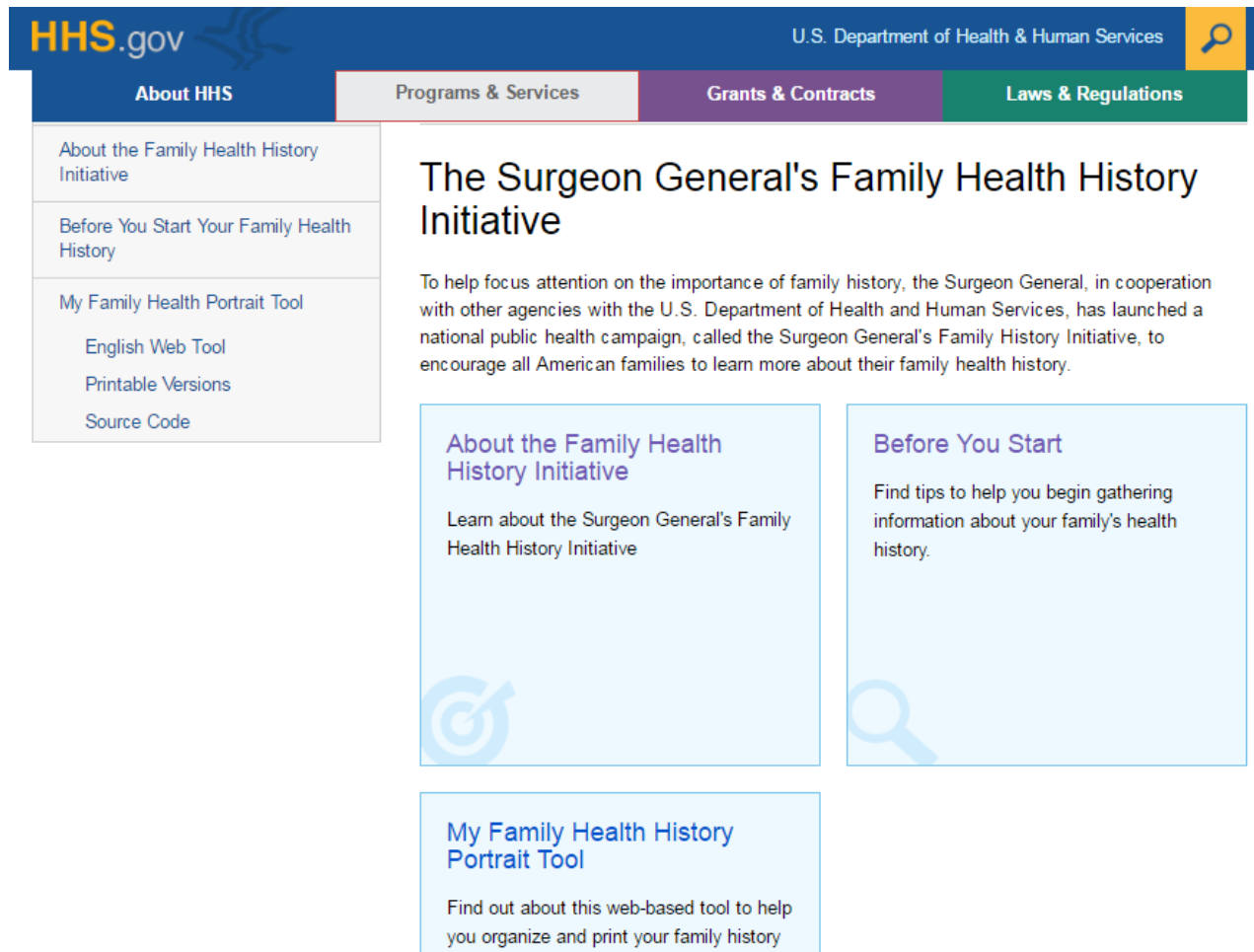
[http://www.nationalacademies.org/hmd/Activities/
PublicHealth/HealthLiteracy/2016-MAR-2.aspx](http://www.nationalacademies.org/hmd/Activities/PublicHealth/HealthLiteracy/2016-MAR-2.aspx)

Tips for communicating to healthcare professionals

- Bring a family member or a friend
- Write down or record the information
- Speak your mind
- Repeat the information as you understand it
- Ask questions (who, what, where, why, how)
- Ask for more information (website, printed handout, a library)

My Family Health Portrait

U.S. Surgeon General



The screenshot shows the HHS.gov website with a blue header. The header includes the HHS.gov logo, the text "U.S. Department of Health & Human Services", and a search icon. Below the header is a navigation bar with four tabs: "About HHS" (selected), "Programs & Services", "Grants & Contracts", and "Laws & Regulations".

Under the "About HHS" tab, there is a sidebar with the following links:

- About the Family Health History Initiative
- Before You Start Your Family Health History
- My Family Health Portrait Tool
 - English Web Tool
 - Printable Versions
 - Source Code

The main content area is titled "The Surgeon General's Family Health History Initiative". Below the title is a paragraph: "To help focus attention on the importance of family history, the Surgeon General, in cooperation with other agencies with the U.S. Department of Health and Human Services, has launched a national public health campaign, called the Surgeon General's Family History Initiative, to encourage all American families to learn more about their family health history."

Below the paragraph are three boxes:

- About the Family Health History Initiative**
Learn about the Surgeon General's Family Health History Initiative
- Before You Start**
Find tips to help you begin gathering information about your family's health history.
- My Family Health History Portrait Tool**
Find out about this web-based tool to help you organize and print your family history

Surgeon General's Family Health History Initiative:

<http://www.hhs.gov/programs/prevention-and-wellness/family-health-history/index.html>

My Family Health Portrait

**NIH** National Human Genome Research Institute

Search Genome.gov

Español



Research Funding Research at NHGRI Health Education Issues Newsroom Careers About

Home > Health > Genomic Medicine for Patients and the Public > Family History: My Family Health Portrait

Genomic Medicine for Patients and the Public
Community Engagement and Community Health Resources
Family History: My Family Health Portrait
Genetic and Genomic Science and Research
Genomic Medicine and Health Care
Specific Genetic Disorders

Family History


Find out how learning your family's health history can help you discover your genetic heritage and risks, and guide you in making healthy environment and lifestyle choices. Learn about how to obtain and create a family health history.

- **My Family Health Portrait**
- **Family History: Resources and Tools** [cdc.gov]
- **Family Health History Tools** [geneticalliance.org]
- **NIH SeniorHealth: Creating a Family Health History** [nihseniorhealth.gov]

NHGRI My Family Health Portrait <https://www.genome.gov/27527640/family-history-my-family-health-portrait/>

My Family Health History

Enter Personal Information

Help

Your Personal Information

We start the family health history with you. Enter the required personal information and your health history information. At the bottom of the page (you may need to scroll), press the 'Next' button. You will then be asked to tell the system which family members you would like to add to the health history.

* Indicates required information.

*Name:

*Gender: ☒ Male ☐ Female

* Date of Birth:
mm/dd/yyyy

Were you born a twin? ☒ No ☐ Yes - Identical (Same) ☐ Yes - Not Identical (Fraternal)

Were you adopted? ☐ Yes

Height Feet Inches -OR- Centimeters

Weight lbs ▾

Your Health Information

In the list below, select a Disease or Condition (if any) from the dropdown box. Then select the Age at Diagnosis and press the Add button. You may repeat this process as necessary.

Disease or Condition	Age at Diagnosis	Action
<div>Please Select a Disease ▾</div>	<div>Select Age at Diagnosis ▾</div>	<div>Add</div>

Your Family Background Information

Check here if your parents are related to each other in any way other than marriage. ☐

Multiple races and ethnicities may be selected.

Race:

☐ American Indian or Alaska Native ☐ Asian ☐ Black or African-American

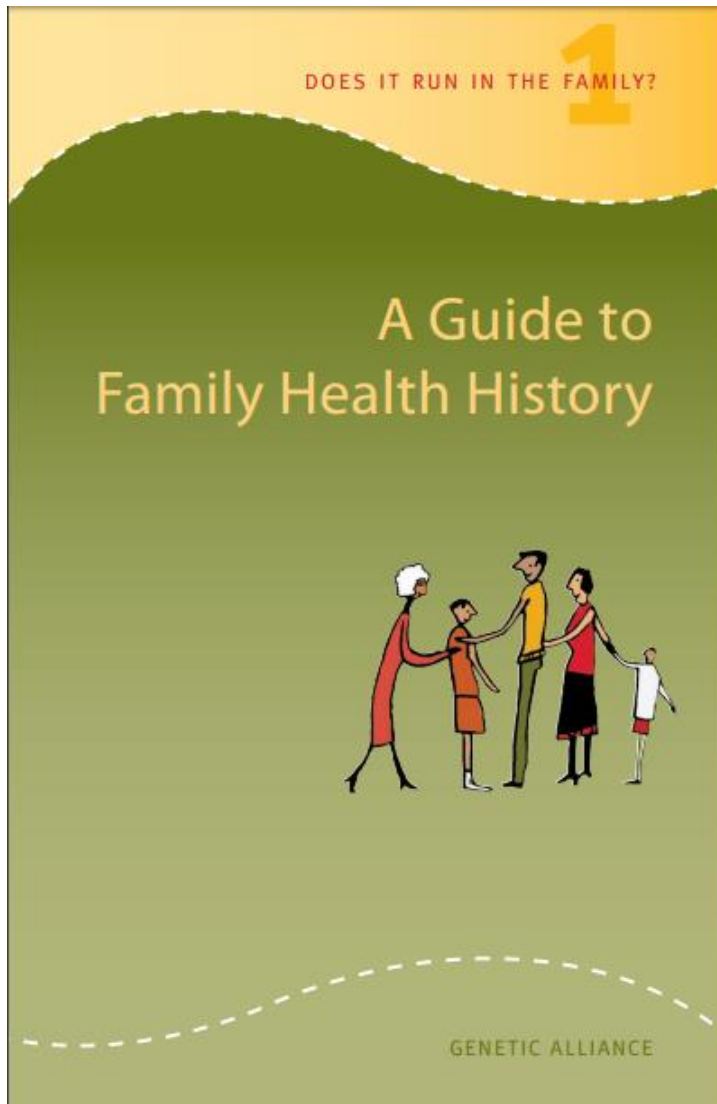
☐ Native Hawaiian or Other Pacific Islander ☐ White

Ethnicity:

☐ Hispanic or Latino ☐ Ashkenazi Jewish ☐ Not Hispanic or Latino

[Why are we asking about Ashkenazi Jewish heritage?](#)

Does It Run In the Family? toolkit



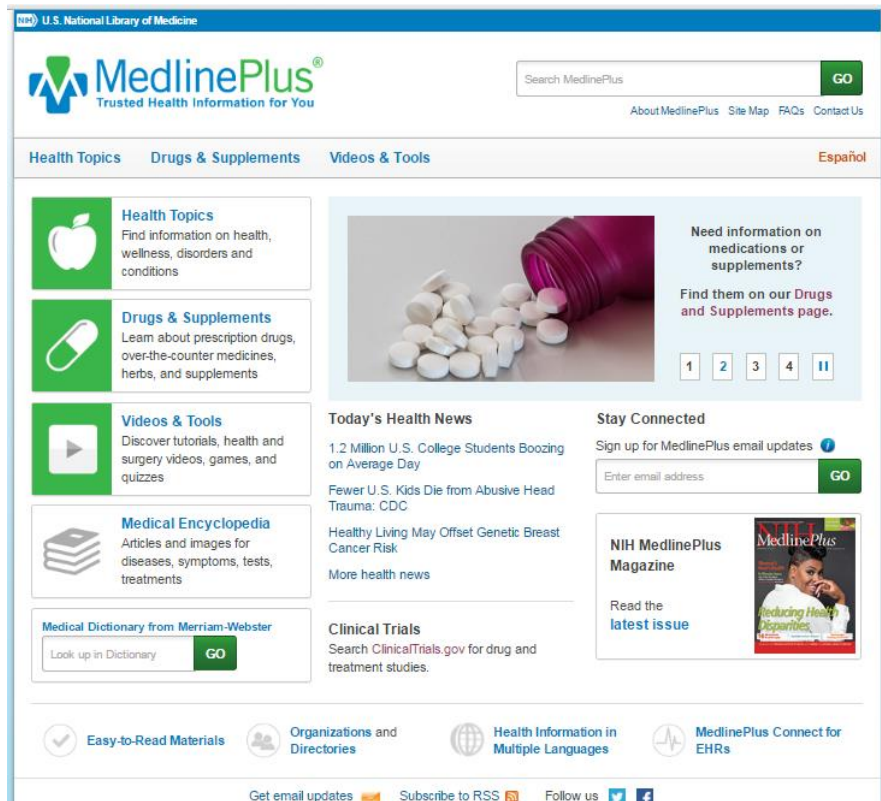
Does it Run In the Family? toolkit

<http://www.geneticalliance.org/sites/default/files/GuideToFHH/GuidetoFHH.pdf>



Consumer Websites

MedlinePlus



- Section: Genetics/Birth Defects
- Health Topic pages:
 - Genetics
 - Genetic testing
 - Genetic counseling
 - Genetic disorders
 - Genes and gene therapy
- text word search

Genetics/Birth Defects

[Health Topics](#)

[Drugs & Supplements](#)

[Videos & Tools](#)

[Home](#) → [Health Topics](#) → [Genetics/Birth Defects](#)

Genetics/Birth Defects

[Abnormalities](#) *see* [Birth Defects](#)

[Achondroplasia](#) *see* [Dwarfism](#)

[Adrenoleukodystrophy](#) *see* [Leukodystrophies](#)

[Alpha-1 Antitrypsin Deficiency](#)

[Amniocentesis](#) *see* [Prenatal Testing](#)

[Anencephaly](#) *see* [Neural Tube Defects](#)

[Arnold-Chiari Malformation](#) *see* [Chiari Malformation](#)

[Ataxia](#) *see* [Friedreich's Ataxia](#)

[Ataxia Telangiectasia](#)

[Birth Defects](#)

[Blood Coagulation Disorders](#) *see* [Hemophilia](#)

[Brain Disorders, Inborn Genetic](#) *see* [Genetic Brain Disorders](#)

[Brain Malformations](#)


[Canavan Disease](#) *see* [Leukodystrophies](#)

[Cephalic Disorders](#) *see* [Brain Malformations](#)

[Cerebral Palsy](#)

[Charcot-Marie-Tooth Disease](#)

MedlinePlus



U.S. National Library of Medicine

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Health TopicsDrugs & SupplementsVideos & Tools

Español

Home → Health Topics → Genetic Disorders

Genetic Disorders

On this page

Basics

- Summary
- Start Here
- Diagnosis and Tests
- Treatments and Therapies

Learn More

- Living With
- Related Issues
- Specifics
- Genetics

See, Play and Learn

- No links available

Research

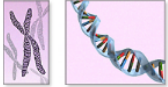

- Statistics and Research
- Clinical Trials
- Journal Articles

Resources

- Reference Desk
- Find an Expert

For You

- Children
- Teenagers
- Women
- Patient Handouts



#ADAM

Get Genetic Disorders updates by email

Enter email address

GO

MEDICAL ENCYCLOPEDIA

[Alström syndrome](#)

[Basal cell nevus syndrome](#)

[Beriberi](#)

[Cystinuria](#)

[Genetics](#)

[Hemochromatosis](#)

Summary

Genes are the building blocks of heredity. They are passed from parent to child. They hold DNA, the instructions for making proteins. Proteins do most of the work in cells. They move molecules from one place to another, build structures, break down toxins, and do many other maintenance jobs.

Sometimes there is a mutation, a change in a gene or genes. The mutation changes the gene's instructions for making a protein, so the protein does not work properly or is missing entirely. This can cause a medical condition called a genetic disorder.

You can inherit a gene mutation from one or both parents. A mutation can also happen during your lifetime.

There are three types of genetic disorders:

- Single gene disorders, when a mutation affects one gene. Cystic fibrosis is an example.

Diagnosis and Tests

- How Are Genetic Conditions Diagnosed? [NHLBI](#) (National Library of Medicine)
- MedlinePlus: Newborn Screening [NHLBI](#) (National Library of Medicine)
Available in Spanish
- Progeria Research Foundation Diagnostic Testing Program (Progeria Research Foundation, Inc.)

Treatments and Therapies

- How Are Genetic Conditions Treated or Managed? [NHLBI](#) (National Library of Medicine)

Living With

- Physical Therapy and Occupational Therapy in Progeria (Progeria Research Foundation, Inc.) - PDF

Related Issues

- Learning about an Undiagnosed Condition in an Adult [NHLBI](#) (National Human Genome Research Institute)

Specifics

- Bloom's Syndrome (Chicago Center for Jewish Genetic Disorders)
- Chromosomal Conditions (March of Dimes Birth Defects Foundation)
Available in Spanish
- Cryopyrin-Associated Autoinflammatory Syndromes (CAPS) - Juvenile (American College of Rheumatology)
- Genetics Home Reference [NHLBI](#) (National Library of Medicine)
- Learning about Poland Anomaly [NHLBI](#) (National Human Genome Research Institute)
- Learning about Progeria [NHLBI](#) (National Human Genome Research Institute)
- Noonan Syndrome (Mayo Foundation for Medical Education and Research)
- Progeria 101/FAQ (Progeria Research Foundation, Inc.)
- Specific Genetic Disorders [NHLBI](#) (National Human Genome Research Institute)
- Triple X Syndrome (Mayo Foundation for Medical Education and Research)
- Williams Syndrome [NHLBI](#) (National Institute of Neurological Disorders and Stroke)

Genetics

- Genetics Home Reference: Genetic Conditions [NHLBI](#) (National Library of Medicine)

Statistics and Research

Cystic Fibrosis
Down Syndrome
Dwarfism
Fragile X Syndrome
Genetic Brain Disorders
Genetic Counseling
Genetic Testing
Hemochromatosis
Leukodystrophies
Osteogenesis Imperfecta
Prader-Willi Syndrome
Rare Diseases
Sickle Cell Anemia

National Institutes of Health

The primary NIH organization for research on *Genetic Disorders* is the [National Institute of Child Health and Human Development](#)

NIH MedlinePlus Magazine

Medical Mysteries: "Thankful They Found a Cause"

Medical Mysteries: "We Feel Deep Compassion for Patients..."

Medical Mysteries: NIH Clinical Center. There's No Other Hospital Like It

Medical Mysteries: NIH Undiagnosed Diseases Program

Promise and Payoff of Rare Diseases Research

MedlinePlus



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Refine by Type

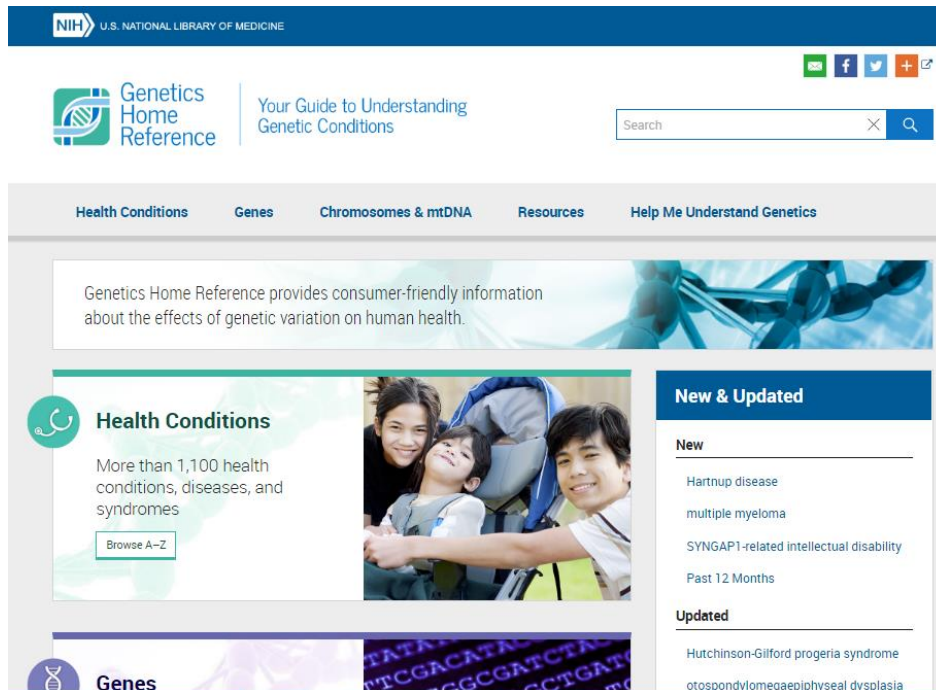
All Results (945)

- ☐ [Health Topics](#) (3)
- ☐ [External Health Links](#) (873)
- ☐ [Drugs and Supplements](#) (10)
- ☐ [Medical Encyclopedia](#) (11)
- ☐ [Videos and Tutorials](#)
- ☐ [News](#) (4)
- ☐ [MedlinePlus Magazine](#) (23)
- ☐ [Other Resources](#) (21)
- ☐ [Multiple Languages](#)

Results 1 - 10 of 945 for **genomics**


1. [Frequently Asked Questions about Genetic and **Genomic** Science](#)  (National Human Genome Research Institute)
... this page Frequently Asked Questions About Genetic and **Genomic** Science What are genetics and **genomics**? Why are ... **genomic** technologies? Additional Resources What are genetics and **genomics**? Genetics is a term that refers to the ...
<https://www.genome.gov/19016904> - External Health Links
2. [Genomics and Health Impact Update](#) (Centers for Disease Control and Prevention)
... Knowledge Base. What's New in the Public Health **Genomics** Knowledge Base New Implementation Tools Alzheimer's Disease Cardiomyopathy HIV/AIDS About the **Genomics** & Health Impact Update The Office of Public Health ...
www.cdc.gov/genomics/update/current.htm - External Health Links
3. [Brief Guide to **Genomics**: DNA, Genes and Genomes](#)  (National Human Genome Research Institute)
... Breve guía de genómica A Brief Guide to **Genomics** DNA, Genes and Genomes Deoxyribonucleic acid (DNA) is ... genetic basis for health and disease. Implications of **Genomics** for Medical Science Virtually every human ailment has ...
<https://www.genome.gov/18016863> - External Health Links
4. [Genomic Testing](#) (Centers for Disease Control and Prevention)

Genetics Home Reference




- Health conditions
- Genes
- Chromosomes and DNA
- Resources
- Genetic handbook

Genetics Home Reference



Health ConditionsGenesChromosomes & mtDNAResourcesHelp Me Understand Genetics



Health Conditions

Explore the signs and symptoms, frequency, genetic cause, and inheritance pattern of various conditions, diseases, and syndromes.

Browse by Category

Browse by First Letter

0-9

A

B

C

D

E

F

G

H

I

J

K

L

M

N

O

P

Q

R

S

T

U

V

W

X

Y

Z

A-[aliphilipoprotein Neuropathy](#), see [Tangier disease](#)
A-T, see [ataxia-telangiectasia](#)
AAA, see [triple A syndrome](#)
AAA syndrome, see [triple A syndrome](#)
AADC deficiency, see [aromatic L-amino acid decarboxylase deficiency](#)
Aarskog syndrome, see [Aarskog-Scott syndrome](#)
[Aarskog-Scott syndrome](#)
AAS, see [Aarskog-Scott syndrome](#)
AASA dehydrogenase deficiency, see [pyridoxine-dependent epilepsy](#)
Aase syndrome, see [Diamond-Blackfan anemia](#)
Aase-Smith syndrome II, see [Diamond-Blackfan anemia](#)
AAT, see [alpha-1 antitrypsin deficiency](#)

Learn More about Health Conditions


What does it mean if a disorder seems to run in my family?

What are the different ways in which a genetic condition can be inherited?

What are complex or multifactorial disorders?

What does it mean to have a genetic predisposition to a disease?


Genetics Home Reference

Health ConditionsGenesChromosomes & mtDNAResourcesHelp Me Understand Genetics

breast cancer

Print AllOpen AllClose All

- Description
- Frequency
- Genetic Changes
- Inheritance Pattern
- Diagnosis & Management
- Other Names for This Condition
- Additional Information & Resources
- Sources for This Page
- Images

Health ConditionsGenesChromosomes & mtDNAResourcesHelp Me Understand Genetics

breast cancer

Print AllOpen AllClose All

- Description
- Frequency
- Genetic Changes
- ▼ Inheritance Pattern

Most cases of breast cancer are not caused by inherited genetic factors. These cancers are associated with somatic mutations in breast cells that are acquired during a person's lifetime, and they do not cluster in families.

In hereditary breast cancer, the way that cancer risk is inherited depends on the gene involved. For example, mutations in the *BRCA1* and *BRCA2* genes are inherited in an **autosomal dominant pattern**, which means one copy of the altered gene in each cell is sufficient to increase a person's chance of developing cancer. Although breast cancer is more common in women than in men, the mutated gene can be inherited from either the mother or the father.

In the other syndromes discussed above, the gene mutations that increase cancer risk also have an autosomal dominant pattern of inheritance. It is important to note that people inherit an increased likelihood of developing cancer, not the disease itself. Not all people who inherit mutations in these genes will ultimately develop cancer.

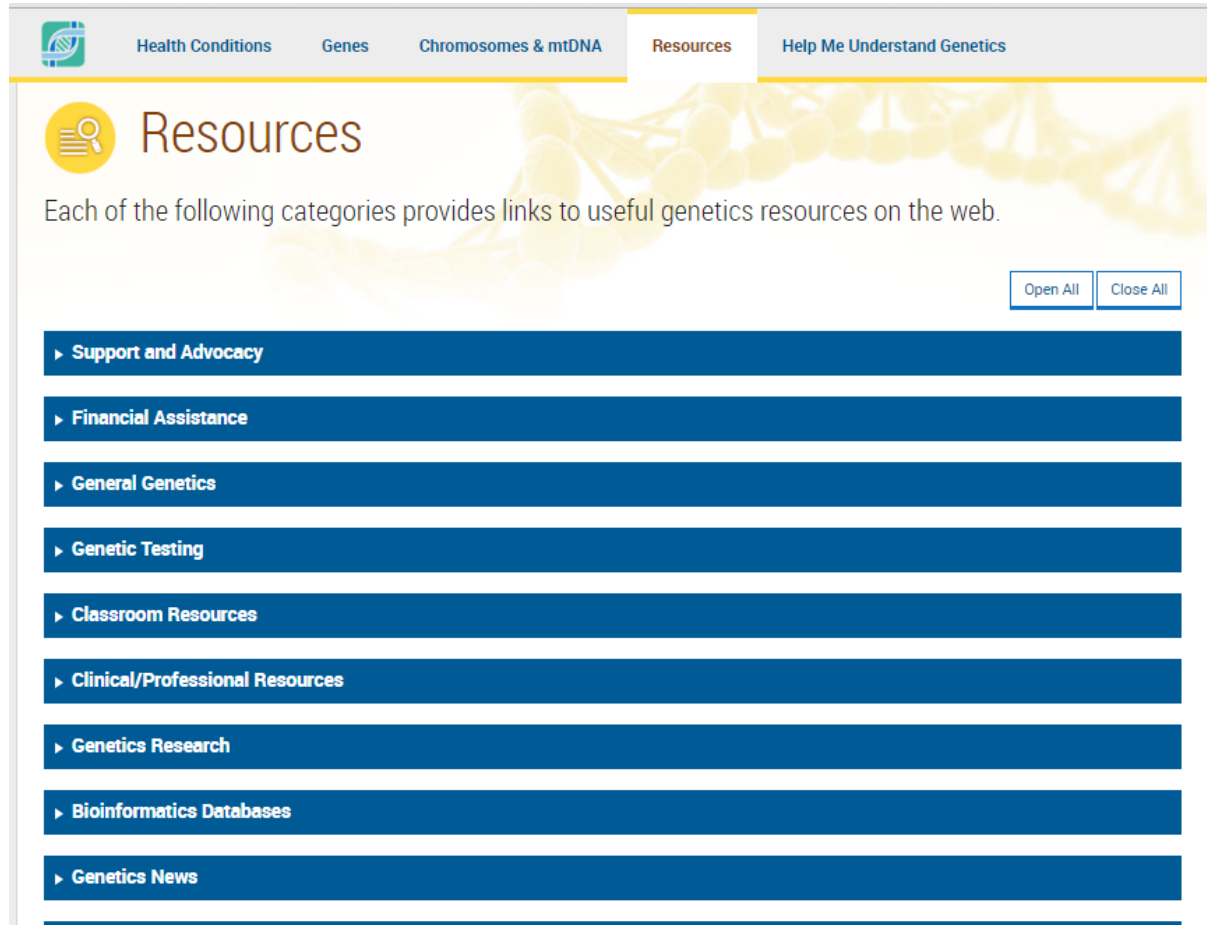
In many cases of breast cancer that clusters in families, the genetic basis for the disease and the mechanism of inheritance are unclear.

Related Information

- [What does it mean if a disorder seems to run in my family?](#)
- [What are the different ways in which a genetic condition can be inherited?](#)
- More about [Inheriting Genetic Conditions](#)

- Diagnosis & Management

Genetics Home Reference



The screenshot shows the 'Resources' page of the Genetics Home Reference website. The page has a navigation bar at the top with links to 'Health Conditions', 'Genes', 'Chromosomes & mtDNA', 'Resources' (which is highlighted), and 'Help Me Understand Genetics'. Below the navigation bar, the 'Resources' section is titled with a magnifying glass icon and the word 'Resources'. A paragraph states: 'Each of the following categories provides links to useful genetics resources on the web.' To the right of this paragraph are two buttons: 'Open All' and 'Close All'. Below this text is a list of ten resource categories, each in a blue bar with a right-pointing arrow: 'Support and Advocacy', 'Financial Assistance', 'General Genetics', 'Genetic Testing', 'Classroom Resources', 'Clinical/Professional Resources', 'Genetics Research', 'Bioinformatics Databases', and 'Genetics News'.

Health Conditions Genes Chromosomes & mtDNA **Resources** Help Me Understand Genetics


Resources


Each of the following categories provides links to useful genetics resources on the web.

[Open All](#) [Close All](#)

- ▶ Support and Advocacy
- ▶ Financial Assistance
- ▶ General Genetics
- ▶ Genetic Testing
- ▶ Classroom Resources
- ▶ Clinical/Professional Resources
- ▶ Genetics Research
- ▶ Bioinformatics Databases
- ▶ Genetics News

Genetics Home Reference

 [Health Conditions](#) [Genes](#) [Chromosomes & mtDNA](#) [Resources](#) [Help Me Understand Genetics](#)



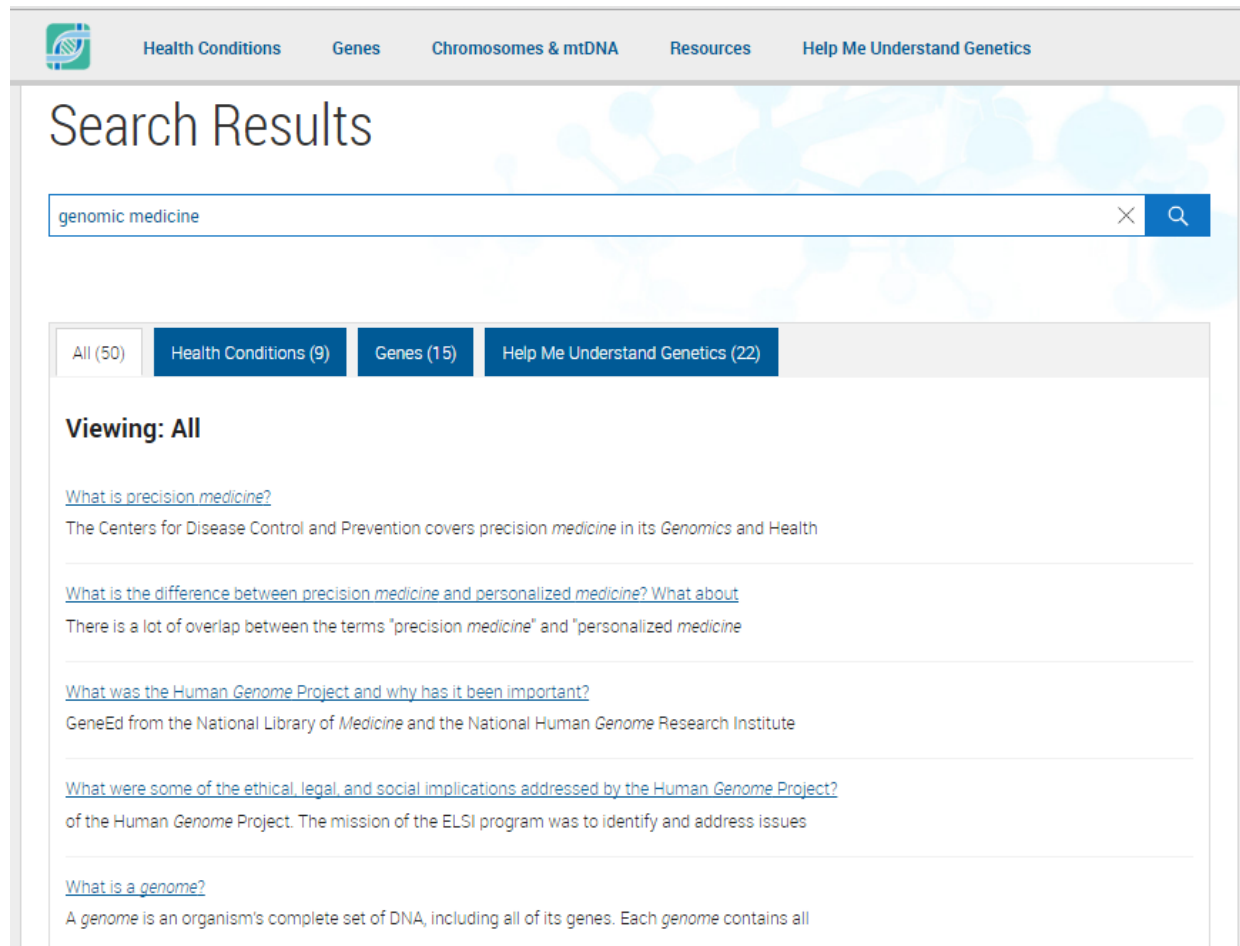
Help Me Understand Genetics

Help Me Understand Genetics provides an introduction to fundamental topics related to human genetics, including illustrations and basic explanations of genetics concepts.

[Download Book \(6MB\)](#) [Open All](#) [Close All](#)

- ▶ **Cells and DNA**
- ▶ **Mutations and Health**
- ▶ **How Genes Work**
- ▶ **Gene Families**
- ▶ **Inheriting Genetic Conditions**
- ▶ **Genetics and Human Traits**
- ▶ **Genetic Consultation**
- ▶ **Genetic Testing**

Genetics Home Reference



The screenshot shows the Genetics Home Reference website. At the top is a navigation bar with a logo and links for Health Conditions, Genes, Chromosomes & mtDNA, Resources, and Help Me Understand Genetics. Below this is a search bar containing the text 'genomic medicine'. The search results are displayed under the heading 'Search Results'. A filter bar shows 'All (50)' selected, with other options for 'Health Conditions (9)', 'Genes (15)', and 'Help Me Understand Genetics (22)'. The results are listed under the heading 'Viewing: All'. The first result is titled 'What is precision medicine?' and is from the Centers for Disease Control and Prevention. The second result is titled 'What is the difference between precision medicine and personalized medicine? What about' and discusses the overlap between the terms. The third result is titled 'What was the Human Genome Project and why has it been important?' and is from GeneEd. The fourth result is titled 'What were some of the ethical, legal, and social implications addressed by the Human Genome Project?' and discusses the ELSI program. The fifth result is titled 'What is a genome?' and defines the term.

Health Conditions Genes Chromosomes & mtDNA Resources Help Me Understand Genetics

Search Results

genomic medicine

All (50) Health Conditions (9) Genes (15) Help Me Understand Genetics (22)

Viewing: All

[What is precision medicine?](#)
The Centers for Disease Control and Prevention covers *precision medicine* in its *Genomics* and Health

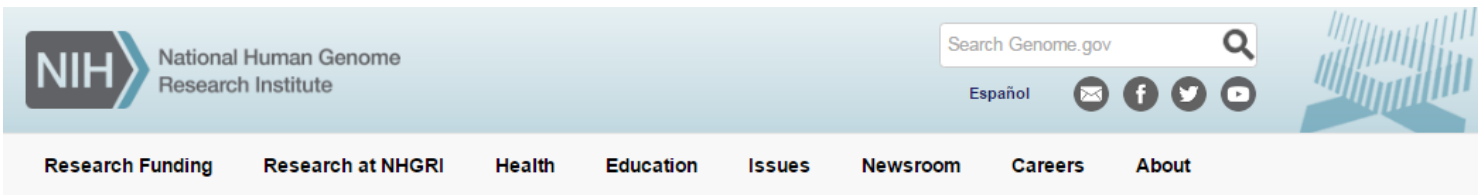
[What is the difference between precision medicine and personalized medicine? What about](#)
There is a lot of overlap between the terms "*precision medicine*" and "*personalized medicine*"

[What was the Human Genome Project and why has it been important?](#)
GeneEd from the National Library of *Medicine* and the National Human *Genome* Research Institute

[What were some of the ethical, legal, and social implications addressed by the Human Genome Project?](#)
of the Human *Genome* Project. The mission of the ELSI program was to identify and address issues

[What is a genome?](#)
A *genome* is an organism's complete set of DNA, including all of its genes. Each *genome* contains all

NIH National Human Genome Research Institute



Health

Information about genetics and genomics, rare diseases, patient care and more

For Patients and the Public



Detailed information about genetic disorders, background on genetic and genomic science, pharmacogenomics, family health history tool and online health resources

- › Community Engagement and Community Health
- › Family History
- › Genetics & Genomics Science & Research
- › Genetic & Rare Diseases Information Center
- › Genomic Medicine and Health Care
- › Online Health and Support Resources
- › Specific Genetic Disorders

For Health Professionals



Genetics and genomics information related to patient management, education, NIH and NHGRI research and ethical, legal and social issues

- › Competency & Curricular Resources
- › Genetics 101
- › Genomic Medicine and Health Care
- › Inter-Society Coordinating Committee (ISCC)
- › New Horizons and Research
- › Patient Management
- › Policy and Ethics Issues

Highlights

NIH awards \$55 million to build million-person precision medicine study



Bethesda, Md., Thurs., July 7, 2016 - The U.S. Food and Drug Administration (FDA) has announced two draft guidances to support President Obama's Precision Medicine Initiative. The guidances will help provide oversight for tests based on next generation sequencing, a technology that examines a person's DNA to detect medically important differences in genomic make-up that could increase the risk for disease.

See Also

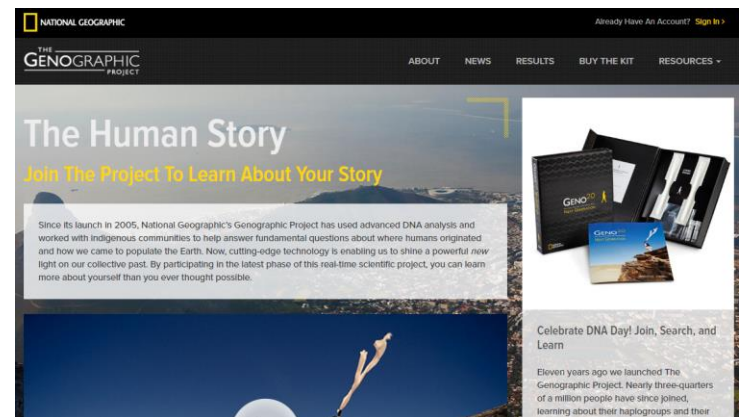
GenomeTV
Genomic Healthcare Branch
Fact Sheets
Genetic Education Resources for Teachers
[All About the Human Genome Project](#)



Direct to Consumer Testing

Genetic Testing

Direct to Consumer Testing



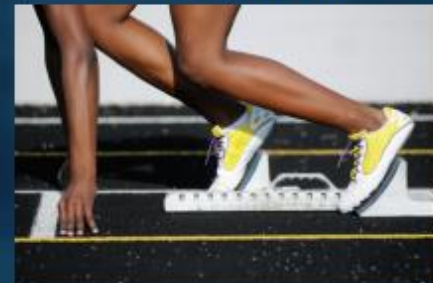
Genomic Testing- Athletic Ability

- ❑ Over 36 companies marketing genetic tests
- ❑ Endurance and power
- ❑ Poor quality control
- ❑ Targeted to coaches and parents
- ❑ Individuals also wanting to focus training

Achieve
your full potential.



Increase
your athletic performance.



Harness
your natural ability through
personalized genomics.



www.GenomicExpress.com

Genomic Testing- Consensus Statement

Consensus statement



OPEN ACCESS

Direct-to-consumer genetic testing for predicting sports performance and talent identification: Consensus statement

Nick Webborn,¹ Alun Williams,² Mike McNamee,³ Claude Bouchard,⁴ Yannis Pitsiladis,⁵ Ildus Ahmetov,⁶ Euan Ashley,⁷ Nuala Byrne,⁸ Silvia Camporesi,⁹ Malcolm Collins,¹⁰ Paul Dijkstra,¹¹ Nir Eynon,¹² Noriyuki Fuku,¹³ Fleur C Garton,¹⁴ Nils Hoppe,¹⁵ Søren Holm,¹⁶ Jane Kaye,¹⁷ Vassilis Klissouras,¹⁸ Alejandro Lucia,¹⁹ Kamiel Maase,²⁰ Colin Moran,²¹ Kathryn N North,¹⁴ Fabio Pigozzi,²² Guan Wang⁵

► Additional material is published online only. To view please visit the journal online (<http://dx.doi.org/10.1136/bjsports-2015-095343>).

For numbered affiliations see end of article.

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Accepted 25 September 2015

ABSTRACT

The general consensus among sport and exercise genetics researchers is that genetic tests have no role to play in talent identification or the individualised prescription of training to maximise performance. Despite the lack of evidence, recent years have witnessed the rise of an emerging market of direct-to-consumer marketing (DTC) tests that claim to be able to identify children's athletic talents. Targeted consumers include mainly coaches and parents. There is concern among the scientific community that the current level of knowledge is being misrepresented for commercial purposes. There remains a lack of universally accepted guidelines and legislation for DTC testing in relation to all forms of genetic testing and not just for talent identification. There is concern over the lack of clarity of information over which specific genes or variants are being tested and the almost universal lack of appropriate genetic counselling for the interpretation of the genetic data to consumers. Furthermore independent studies have identified issues relating to quality control by DTC laboratories with different results being reported from

of the evidence in relation to genetic testing and the limitations of current knowledge. This article reviews the issues around the currently available evidence behind the genetic testing, comments on the ethical considerations and makes recommendations about such tests.

STATEMENT ON BACKGROUND TO THE CONSENSUS PROCESS

A group of world experts in the field of genomics, exercise, sport performance, disease, injury and antidoping gathered with the International Federation of Sports Medicine (FIMS) Scientific Commission for a symposium to discuss the current state of knowledge and to share ideas. One key concern was the misuse of research evidence and the misinformation about genetic testing, particularly when marketed directly to the public, coaches or parents. This is known as DTC testing for the purpose of talent identification and to assess potential for future sports performance. There have been

Concerns

- Privacy
- Legality
- Who has access?
- How useful now?
- What all is being done now and in the future with the information?
- Unexpected surprises?
- Test results can vary among companies
- Validity of tests
- No counseling provided
- Who can get the testing?

Benefits



- ❑ Learn more about own health
- ❑ More effective medical treatments
- ❑ Learn more about ethnicity and family history
- ❑ Bring awareness to family health issues for future generations
- ❑ Motivation to work on health habits
- ❑ Encourages patient engagement
- ❑ Contributing to advancement of healthcare and science
- ❑ Moral obligation



□ (example of test results from 23andMe)

Genetics Home Reference

NIH U.S. NATIONAL LIBRARY OF MEDICINE



Your Guide to Understanding Genetic Conditions



Health Conditions

Genes

Chromosomes & mtDNA

Resources

Help Me Understand Genetics

Home

Help Me Understand Genetics

Genetic Testing


What is direct-to-consumer genetic testing?


What is direct-to-consumer genetic testing?

Traditionally, genetic tests have been available only through healthcare providers such as physicians, nurse practitioners, and genetic counselors. Healthcare providers order the appropriate test from a laboratory, collect and send the samples, and interpret the test results. Direct-to-consumer genetic testing refers to genetic tests that are marketed directly to consumers via television, print advertisements, or the Internet. This form of testing, which is also known as at-home genetic testing, provides access to a person's genetic information without necessarily involving a doctor or insurance company in the process.

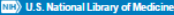
If a consumer chooses to purchase a genetic test directly, the test kit is mailed to the consumer instead of being ordered through a doctor's office. The test typically involves collecting a DNA sample at home, often by swabbing the inside of the cheek, and mailing the sample back to the laboratory. In some cases, the person must visit a health clinic to have blood drawn. Consumers are notified of their results by mail or over the telephone, or the results are posted online. In some cases, a genetic counselor or other healthcare provider is available to explain the results and answer questions. The price for this type of at-home genetic testing ranges from several hundred dollars to more than a thousand dollars.

For more information about direct-to-consumer genetic testing:

The American College of Medical Genetics, which is a national association of doctors specializing in genetics, has issued [a statement on direct-to-consumer genetic testing](#) .

The American Society of Human Genetics, a professional membership organization for specialists in genetics, has also issued [a statement on direct-to-consumer genetic testing in the United States](#) .

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Genetic Testing

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Research <ul style="list-style-type: none">Statistics and ResearchClinical TrialsJournal Articles	Resources <ul style="list-style-type: none">Reference DeskFind an Expert	For You <ul style="list-style-type: none">Patient Handouts

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Buccal smear
Genetic testing and your cancer risk

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Birth Defects

Summary

Genetic tests are tests on blood and other tissue to find **genetic disorders**. Over 2000 tests are available. Doctors use genetic tests for several reasons. These include

- Finding genetic diseases in unborn babies
- Finding out if people carry a gene for a disease and might pass it on to their children
- Screening embryos for disease
- Testing for genetic diseases in adults before they cause symptoms
- Making a diagnosis in a person who has disease symptoms
- Figuring out the type or dose of a medicine that is best for a certain person

People have many different reasons for being tested or not being tested. For some, it is important to know whether a disease can be prevented or treated if a test is positive. In some cases, there is no treatment. But test results might help a person make life decisions, such as family planning or insurance coverage. A **genetic counselor** can provide information about the pros and cons of testing.

NLM: National Human Genome Research Institute

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Direct-to-Consumer Genetic Tests


Could a simple medical test tell you if you are likely to get a particular disease? Could it evaluate your health risks and even suggest a specific treatment? Could you take this test in the privacy of your home, without a doctor's prescription or guidance?

Some companies say genetic testing can do all this and more. They claim that direct-to-consumer (DTC) genetic testing can screen for diseases and provide a basis for choosing a particular diet, dietary supplement, lifestyle change, or medication. These companies primarily sell their tests online and through multi-level marketing networks.

The Federal Trade Commission (FTC) wants you to know the facts about the DTC marketing of genetic tests.

[View this page in español](#)

Related Items


Anatomy of a Cancer Treatment Scam

- Dietary Supplements
- Miracle Health Claims
- Cancer Treatment Scams

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[→ Interpreting the Results](#)
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[→ If You're Considering a DTC Genetic Test](#)
[→ For More Information](#)

American College of Medical Genetics and Genomics

Folic Acid

Folic Acid and Neural Tube Defects	2010, ACMG	2010 Genet Med 13:6:593-596
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Genetic Discrimination

Points to Consider in Preventing Unfair Discrimination Based on Genetic Disease Risk: A Position Statement of the American College of Medical Genetics and Genomics	2001, ACMG Reaffirmed 2005	2001 Genet Med 3:6: 436-437
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Genetic Services

Clinical utility of genetic and genomic services: a position statement of the American College of Medical Genetics and Genomics	2015, ACMG	2015 Genet Med 7:6:505-507
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Genetic Testing

ACMG Revised Position Statement on Direct-to-Consumer Genetic Testing	2015, ACMG	2016 Genet Med 18:2:207-208
ACMG position statement on prenatal/ preconception expanded carrier screening	2013, ACMG	2013 Genet Med 15:6:482-483
Technical report: ethical and policy issues in genetic testing and screening of children	2013, ACMG/AAP	2013 Genet Med 15:3:234-245
Risk categorization for oversight of laboratory-developed tests for inherited conditions	2013, ACMG	2013 Genet Med 15:4:314-5
Genetic Testing in Adoption (ACMG/ASHG)	2000, ASHG	2000 Am J Hum Genet 66:761-767

Genomic Sequencing

ACMG Policy Statement: Updated Recommendations Regarding Analysis and Reporting of Secondary Findings in Clinical Genome-Scale Sequencing	2015, ACMG	2015 Genet Med 17:1:68-69
ACMG Recommendations for reporting of incidental findings in clinical exome and	2013,	2013 Genet

© American College of Medical Genetics and Genomics

ACMG STATEMENT | Genetics in Medicine

Direct-to-consumer genetic testing: a revised position statement of the American College of Medical Genetics and Genomics

ACMG Board of Directors¹

Disclaimer: These recommendations are designed primarily as an educational resource for medical geneticists and other health-care providers to help them provide quality medical genetics services. Adherence to these recommendations does not necessarily assure a successful medical outcome. These recommendations should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. In determining the propriety of any specific procedure or test, geneticists and other

clinicians should apply their own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. It may be prudent, however, to document in the patient's record the rationale for any significant deviation from the recommendations.

Genet Med advance online publication 17 December 2015

Key Words: consumer; direct-to-consumer; genetic testing; self-testing; OTC

With ongoing genetic discoveries and improvements in technology, more genetic tests are available than ever before. Along with greater availability has come increased consumer demand for genetic tests and expansion of direct-to-consumer testing. The American College of Medical Genetics and Genomics (ACMG) has revised its 2008 e-publication regarding this issue (ACMG Statement on Direct-to-Consumer Genetic Testing, retired; available by request to acmg@acmg.net) and believes that it is critical for the public to realize that genetic testing is only one part of a complex process that includes genetic risk

A genetics expert such as a certified medical geneticist or genetic counselor should be available to help the consumer determine, for example, whether a genetic test should be performed and how to interpret test results in light of personal and family history. A board-certified genetic counselor can help facilitate this process by providing information about the test and helping to explain test results. A number of risks can be reduced if a board-certified genetics professional is involved in genetic testing, including inadequate or lack of informed consent

ACMG <https://www.acmg.net/>



Ethics and Privacy

Societal Concerns

- Who should have access to personal genetic information, and how will it be used?
- Who owns and controls genetic information?
- How does personal genetic information affect an individual and society's perceptions of that individual?
- What are the larger societal issues raised by new reproductive technologies?
- How will genetic tests be evaluated and regulated for accuracy, reliability and utility?
- How do we prepare healthcare professionals and the public?
- What is considered acceptable diversity?
- Where is the line between medical treatment and enhancement?
- Should testing be performed when no treatment is available?

GINA

GINA

GENETIC INFORMATION
NONDISCRIMINATION ACT

[About](#) | [Contact](#)

Genetic Information

What is genetic information and why is it important?

GINA & Health Insurance

What are GINA's health insurance protections?

GINA & Employment

What are GINA's employment protections?

What is GINA?

The Genetic Information Nondiscrimination Act of 2008 (GINA) is a federal law that protects individuals from genetic discrimination in health insurance and employment. Genetic discrimination is the misuse of genetic information. This resource provides an introduction to GINA and its protections in health insurance and employment. It includes answers to common questions and examples to help you learn. Choose from one of the boxes to the left to begin!

✉ Have questions, comments or suggestions? [Send us a note.](#)

🖨 [Click here](#) for a printer friendly version.

✉ For healthcare provider resources [click here.](#)

🖨 [Click here](#) for the GINA & You Information Sheet

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Catalog item

Choosing Wisely

<http://consumerhealthchoices.org/catalog/making-smart-decisions-about-genetic-testing-acmg/>

Making Smart Decisions About Genetic Testing (ACMG)

Campaign Series: Choosing Wisely

Medical Category: Tests

Article Type: Advice

Language: Plain English, Spanish

Affiliation: American College of Medical Genetics and Genomics

Format: HTML, PDF

Most recent update: 10/07/2015



Sometimes a genetic test is not the best way to find an inherited condition or disease risk. A routine blood test or procedure might be just as good. And it might be less costly and more easily available.

Files to download

Plain English: [Making Smart Decisions About Genetic Testing \(ACMG\) →](#)

Spanish: [Cómo tomar decisiones inteligentes sobre las pruebas genéticas →](#)

[See webpage version](#)

Webpage file: [Making Smart Decisions About Genetic Testing →](#)

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Webpage file: [Cómo tomar decisiones inteligentes sobre las pruebas genéticas →](#)

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ACMG NEWS For Immediate Release

Contact: Kathy Beal, MBA
ACMG Media Relations
301-238-4582, kbeal@acmg.net

ACMG Provides Recommendations on Genetic Testing Through the Choosing Wisely® Campaign

Goal is to encourage clinician and patient conversations about appropriate genetic testing

Bethesda, MD -- July 10, 2015- The American College of Medical Genetics and Genomics (ACMG) has released a list of five things patients and providers should discuss regarding specific genetic tests as part of *Choosing Wisely*, an initiative of the ABIM Foundation. The just-released list identifies five evidence-based recommendations that can support conversations between patients and clinicians regarding genetic testing. To date, more than 100 national and state medical specialty societies, regional health collaboratives and consumer partners have joined the *Choosing Wisely* campaign about appropriate care.

The list provided by ACMG provides evidence-based recommendations to help all clinicians and patients to have conversations about making wise choices related to genetic testing. The lists were developed over the past year with careful consideration of the latest evidence, expert opinions and research.

ACMG

https://www.acmg.net/docs/ACMG_ChoosingWisely_Final.pdf

NIH National Human Genome Research Institute



Issues in Genetics

Policy, legal and ethical issues in genetic research



Coverage and Reimbursement of Genetic Tests
Information about insurance coverage for genetic testing



Human Subjects Research
Human subject participation for biomedical, clinical and social-behavioral research



Genetic Discrimination
How Americans are protected from discrimination based on their genetics



Regulation of Genetic Tests
How the federal government regulates genetic tests.



Privacy in Genomics
How best to ensure that genomic information remains private



Informed Consent
The rights of participants when consenting to research projects



Intellectual Property and Genomics
Can a gene be patented?



Genetics and Public Policy Fellowship
A fellowship for genetics professionals interested in public policy



Genome Statute and Legislation Database
A database of state statutes and bills from 2007-2016 U.S. state legislative sessions

Highlights

FDA requests comments on draft guidance for Precision Medicine Initiative



The U.S. Food and Drug Administration (FDA) has announced two draft guidances to support President Obama's Precision Medicine Initiative. The guidances will help provide oversight for tests based on next generation sequencing.

See Also

Policy and Program Analysis Branch
Staff Contact Information

Ethical, Legal and Social Implications Research Program
NHGRI's Extramural Research Program

GenomeTV

American Academy of Pediatrics American College of Medical Genetics and Genomics

Pediatrics

March 2013, VOLUME 131 / ISSUE 3

From the American Academy of Pediatrics

Policy Statement

AAP

<http://pediatrics.aappublications.org/content/131/3/620>

Ethical and Policy Issues in Genetic Testing and Screening of Children

COMMITTEE ON BIOETHICS, COMMITTEE ON GENETICS, AND, THE AMERICAN COLLEGE OF MEDICAL GENETICS AND, GENOMICS SOCIAL, ETHICAL, AND LEGAL ISSUES COMMITTEE

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Abstract

The genetic testing and genetic screening of children are commonplace. Decisions about whether to offer genetic testing and screening should be driven by the best interest of the child. The growing literature on the psychosocial and clinical effects of such testing and screening can help inform best practices. This policy statement represents recommendations developed collaboratively by the American Academy of Pediatrics and the American College of Medical Genetics and Genomics with respect to many of the scenarios in which genetic testing and screening can occur.

ACMG

https://www.acmg.net/docs/genetic_testing_in_children_preprint_gim2012176a.pdf

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ACMG POLICY STATEMENT

Genetics
inMedicine

Technical report: ethical and policy issues in genetic testing and screening of children

Laine Friedman Ross, MD, PhD¹⁻², Howard M. Saal, MD³, Karen L. David, MD, MS^{4,6} and Rebecca R. Anderson, JD, MS⁵; and the American Academy of Pediatrics; American College of Medical Genetics and Genomics

The genetic testing and genetic screening of children are commonplace. Decisions about whether to offer genetic testing and screening should be driven by the best interest of the child. The growing literature on the psychosocial and clinical effects of such testing and screening can help inform best practices. This technical report provides ethical justification and empirical data in support of the pro-

posed policy recommendations regarding such practices in a myriad of settings.

Genet Med advance online publication 21 February 2013

Key Words: carrier identification; disclosure; genetic screening; genetic testing; newborn screening; predictive testing

INTRODUCTION

Two major events occurred in the 1950s that forever changed the influence of genetics in medicine: Watson and Crick¹ described the double-helix model of DNA structure in 1953, and in 1956 Tjio and Levan² established that the typical human carries 46 chromosomes. The goal of mapping and sequencing the human genome began in 1990, and a working draft was presented in 2000, with a more complete edition published in 2003.³ Knowledge of genetics and genomics continues to grow rapidly, as does consumer interest in genetic testing. As a result, statements about genetic testing and screening of children in the United States written in the past two decades need to be updated to consider the ethical issues that arise with the new technologies and expanded uses of genetic testing and screen-

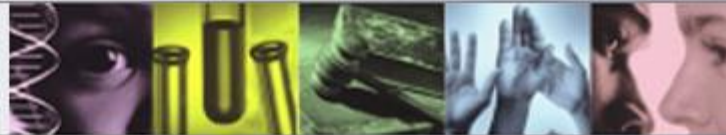
hematologic, and endocrine abnormalities for which early treatment may prevent or reduce morbidity or mortality. Most of the genetic conditions included in the state screening panels are autosomal recessive disorders, and some assays identify heterozygote carriers (e.g., hemoglobinopathies). Future screening may expand to X-linked conditions (e.g., Duchenne muscular dystrophy) and autosomal dominant conditions. In addition, universal newborn hearing screening allows for early identification of both acquired and hereditary hearing loss.

Outside of newborn screening, pediatric genetic testing is much less common. Diagnostic genetic testing may be performed on a child with physical, developmental, or behavioral features consistent with a potential genetic syndrome or for pharmacogenetic drug selection and dosing decisions.

Informing the Public



CENTER FOR
GENETICS AND
SOCIETY



Precision Medicine

“...a bold new research effort to revolutionize how we improve health and treat disease.”

[PMI announcement](https://www.whitehouse.gov/precision-medicine) <https://www.whitehouse.gov/precision-medicine>

Precision Medicine Initiative

Mission statement:

To enable a new era of medicine through research, technology, and policies that empower patients, researchers, and providers to work together toward development of individualized care.



THE PRECISION MEDICINE INITIATIVE



PRECISION MEDICINE INITIATIVE PRINCIPLES STORIES

f t GO TO TOP

[PMI announcement](https://www.whitehouse.gov/precision-medicine) <https://www.whitehouse.gov/precision-medicine>

Precision Medicine is...



- Precision medicine is an emerging approach for disease treatment and prevention that takes into account individual variability in genes, environment, and lifestyle for each person.
- Instead of what treatment is right for this disease it is what treatment is right for this patient.

Precision Medicine Initiative

□ **Near Term goals:**

- Clinical trials focusing on pediatric cancers and drug therapies for adults
- Use of combination therapies
- Overcoming drug resistance

□ **Long Term Goals:**

- Create research cohort of 1 million volunteers
- New model of medicine
 - engage participants
 - responsible data sharing
 - privacy protection
- Advance pharmacogenomics
- Identify new targets for treatment and prevention
- Test if mobile devices encourages healthy behaviors
- Lay scientific foundation for many diseases

Precision Medicine Cohort Program

- Two ways to participate
 1. Through the cohort website
 2. With participating health care provider organization



**PRECISION MEDICINE INITIATIVE®
COHORT PROGRAM**

WHAT IS IT?

Precision medicine is a groundbreaking approach to disease prevention and treatment based on people's individual differences in environment, genes and lifestyle.

The Precision Medicine Initiative® Cohort Program will lay the foundation for using this approach in **clinical practice**.

WHAT ARE THE GOALS?

Engage a group of **1 million or more U.S. research participants** who will share biological samples, genetic data and lifestyle information, all linked to their electronic health records. This data will allow researchers to develop more precise treatments for **many diseases and conditions**.

Pioneer a new model of research that emphasizes **engaged research participants, responsible data sharing and privacy protection**.

Research based on the cohort data will:

- Lay **scientific foundation** for precision medicine
- Help identify new ways to **treat and prevent disease**
- Test whether **mobile devices**, such as phones and tablets, can encourage healthy behaviors
- Help develop the **right drug** for the **right person** at the **right dose**

WHY NOW?

The **time is right** because:

- We have a greater understanding of human genes
- People are more engaged in healthcare and research
- We have the tools to track health information and use large databases
- Research technologies have improved

Follow the Initiative's progress and be one of the first to join this landmark effort.

NIH and Precision Medicine Initiative

PRECISION MEDICINE INITIATIVE COHORT PROGRAM

Precision Medicine Initiative

[Scale and Scope](#)

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NIH awards \$55 million to build million-person precision medicine study



Learn about the key components of the PMI Cohort Program

About the Precision Medicine Initiative Cohort Program

Far too many diseases do not have a proven means of prevention or effective treatments. We must gain better insights into the biological, environmental, and behavioral influences on these diseases to make a difference for the millions of Americans who suffer from them. Precision medicine is an emerging approach for disease treatment and prevention that takes into account individual variability in genes, environment, and lifestyle for each person. While some advances in precision medicine have been made, the practice is not currently in use for most diseases.



Email Updates

Sign up to receive email updates about the Precision Medicine Initiative.

[Sign up for updates](#)

Related Links

[PMI Working Group Final Report](#)

[pdf](#)

[NEJM Perspective: A New Initiative on Precision Medicine](#)

[White House Precision Medicine Web Page](#)

[White House Fact Sheet: President Obama's Precision Medicine Initiative](#)

[Precision Medicine Initiative and Cancer Research](#)

[Precision Medicine Initiative YouTube Channel](#)

[PMI Cohort Program ://www.nih.gov/precision-medicine-initiative-cohort-program](http://www.nih.gov/precision-medicine-initiative-cohort-program)

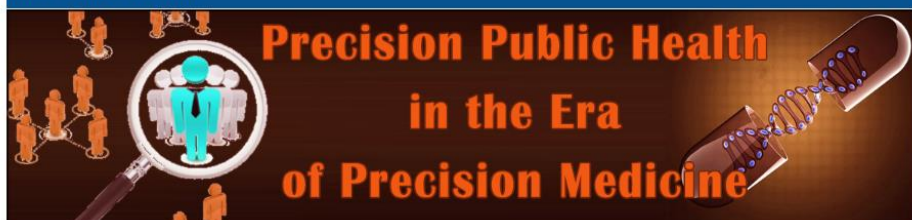
MedlinePlus Magazine- Fall 2015



Health Care Tailored to You



Public Health Genomics



What's New in Public Health Genomics Knowledge Base



ASTHMA



BIG DATA



HEPATITIS

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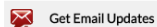
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Precision Medicine and Population Health: Dealing With the Elephant in the Room



In this week's Journal of American Medical Association, we published a point-counterpoint commentary on the impact of precision medicine on population health. Announcement of the news of the US precision medicine initiative has been met with a range of responses from enthusiasm to skepticism about potential benefits, limitations and return on investment. In considering the [Read More >](#)

Posted on August 17, 2016 by Mulin J. Khoury, Director, Office of Public Health Genomics, Centers for Disease Control and Prevention, Atlanta, Georgia, and Sandro Galea, Dean, School of Public Health, Boston University, Boston, Massachusetts

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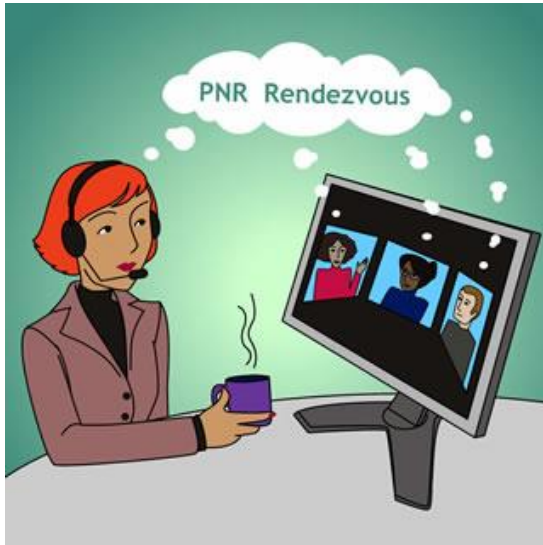
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<http://blogs.cdc.gov/genomics/>

[Five Misconceptions About the Role of Genomics in Public Health](#)

PNR Rendezvous



Adventures in Precision Medicine: A Major Public Research Initiative and its Implications for Healthcare Consumers and Institutions September 21, 2016

Presenter: Malia Fullerton,
Associate Professor of Bioethics and
Humanities at the University of
Washington School of Medicine

Library role



- “Preparing the public to make educated personal and family health decisions in a time of rapidly evolving genetic and genomic knowledge will require new partnerships between the education system, health care systems, the government, community advocacy organizations, consumers and the media.”

Show What You Know!

- The 1000 Genomes Project was undertaken in order to increase the _____ of the genomes represented in public databases.
- What term refers to strategies for determining what treatment is right for an INDIVIDUAL rather than what treatment is recommended for a DISEASE?
- Clinicians are not concerned about all genetic variants – only those that are _____.
- True or False? GINA (Genetic Information Nondiscrimination Act) protects you from life insurance discrimination.
- True or False? A genetic variant may originally be classified as “likely pathogenic” and later classified as “likely benign.”
- What resource would you recommend to consumers who wanted to learn more about a genetic condition?
- What is a good starting place for finding genetic information for clinicians?

Questions?

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Presentation resources

<https://nnlm.gov/pnr/training/presentations>

